SUPPLEMENTAL TABLES

Supplementary Table 1

The DupMasker program (Jiang et al. 2008), a genome annotation tool that uses a library of nonredundant consensus sequences of human segmental duplication, is applied to delineate the order and orientation of duplications within 17q21.31 inversion polymorphism for H1 haplotype (1.169 kb) (Zody et al. 2008). Strand annotations are as follows: SW score = smith-waterman score of the match (complexity-adjusted), perc div. = %substitutions in matching region, perc del. = %deletions (in query sequence relative to subject) in matching region, perc ins. = %insertions (in query sequence relative to subject) in matching region, qry seq = id of query sequence, qry position begin = starting position of match in query sequence, qry position end = ending position of match in query sequence, qry (left) = no. of bases in query sequence past the ending position of match (so 0 means that the match extended all the way to the end of the query sequence), C = "C" match is found on the reverse strand, subj seq = id of the duplicon, subj (left) = The remaining bases in (complement of) subject sequence prior to beginning of the match, subj position end = starting position of match in subject sequence (using top-strand numbering), subj position begin = ending position of match in subject sequence. Green and yellow highlighted sequences indicate the position of Cl3 and Cl8 promoters, respectively. Please note that the library construction for nonredundant consensus sequences of human segmental duplication is based on build35/hg17.

Supplementary Table 2

The DupMasker program (Jiang et al. 2008), a genome annotation tool that uses a library of nonredundant consensus sequences of human segmental duplication, is applied to delineate the order and orientation of duplications within 17q21.31 inversion polymorphism for H2 haplotype (1.481kb) (Zody et al. 2008). Strand annotations are as follows: SW score = smith-waterman score of the match (complexity-adjusted), perc div. = %substitutions in matching region, perc del. = %deletions (in query sequence relative to subject) in matching region, perc ins. = %insertions (in query sequence relative to subject) in matching region, qry seq = id of query sequence, qry position begin = starting position of match in query sequence, qry position end = ending position of match in query sequence, qry (left) = no. of bases in query sequence past the ending position of match (so 0 means that the match extended all the way to the end of the query sequence), C = "C" match is found on the reverse strand, subj seq = id of the duplicon, subj (left) = The remaining bases in (complement of) subject sequence prior to beginning of the match, subj position end = starting position of match in subject sequence (using top-strand numbering), subj position begin = ending position of match in subject sequence. Green and yellow highlighted sequences indicate the position of Cl3 and Cl8 promoters, respectively. Please note that the library construction for nonredundant consensus sequences of human segmental duplication is based on build35/hg17.

Supplementary Table 3

PCR was performed in 20 µl reactions composed of 0.8 µl of a 10 µM dilution of the forward primer and reverse primer, 10 µl of Roche (11636103001) PCR Master Mix. The following PCR conditions were used (A): 1 min at 94^{o}C, followed by 38 cycles at 94^{o}C

for 30 sec, 55°C 30 sec, and 72°C for 30 sec followed by 7 min at 72°C. The following real-time PCR conditions (B) were used: 3 min at 95°C, followed by 40 cycles at 95°C for 15 sec, 55°C 20 sec, and 72°C for 20 sec.